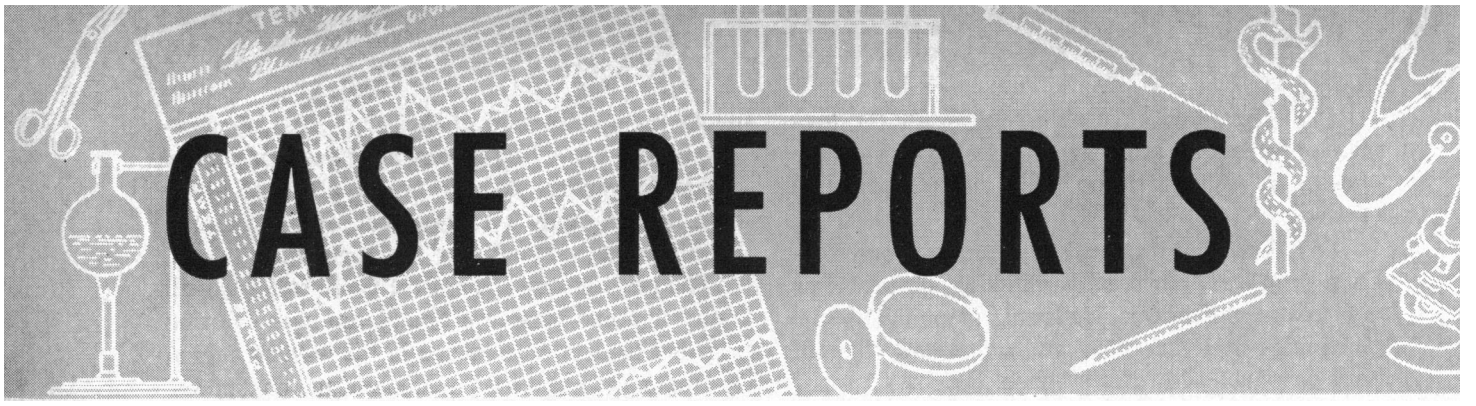

Gynecomastia, Hypospermatogenesis and a Large Y Chromosome in a Man

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UNTIL THE RECENTLY described YY Syndrome,[1,3,7,8,9] characterized by the tall, mentally retarded, and institutionalized or criminal male, the Y chromosome abnormalities were clinically unrecognized.

This report concerns a young adult male with gynecomastia and hypospermatogenesis who has an elongated Y chromosome.

Report of a Case

A 20-year-old male college student was in good health until January 1961, when he noted a slow and painless breast enlargement, greater on the left side than the right. The breast development became a severe emotional problem, for which he sought medical treatment.

The patient described normal male libido.

On physical examination in March 1964 the patient was observed to be well developed, well nourished and of normal proportions. Breast enlargement with palpable glandular tissue was noted on both sides. The nipples were flat. Genitalia and hair distribution were normal.

Blood cell counts and results of urinalysis were within normal limits. X-ray films of the skull and intravenous urograms showed no abnormality. Reaction to a frog test for pregnancy was negative. Results of a 24-hour specimen of urine (1160 ml) were as follows:

Pregnanediol, 2.3 mg; pituitary gonadotropin (follicle stimulating hormone) less than 6 units; 17 ketosteroids, 12.8 mg in 23 hours; 17 ketogenic steroids 17.4 mg in 14 hours.

Microscopic examination of a testicular biopsy specimen showed pronounced disorganization of spermatogenesis and hypospermatogenesis.

The patient was admitted to the hospital where simple mastectomy was performed, approximately 200 gm of tissue being removed from each breast. Microscopic examination of the operative specimens showed increased collagenous connective tissue surrounding nests of increased mammary ducts. The ducts were lined by several layers of epithelial cells which were proliferating focally. It was the pathologist's impression that the slight ductal hyperplasia was consistent with a diagnosis of gynecomastia.

At this time, cytogenetic studies were completed. A buccal smear revealed 100 per cent chromatin-negative cells.

Preparations of the patient's chromosomes were obtained from peripheral blood. The leukocytes were cultured in Ham's F10 media. After three days, the cells were harvested, treated with colchicine, placed in hypotonic solution, fixed and stained. The figures were analyzed as described by Patau.[6] Thirty-five figures were studied; 30 of them had 46 chromosomes; five had a non-modal number, but an inconstant pattern, and were believed to be non-modal due to broken cells. An unusually large acrocentric chromosome (see Figure 1) was present in all figures.

Discussion

The mass of the large Y chromosomes found in

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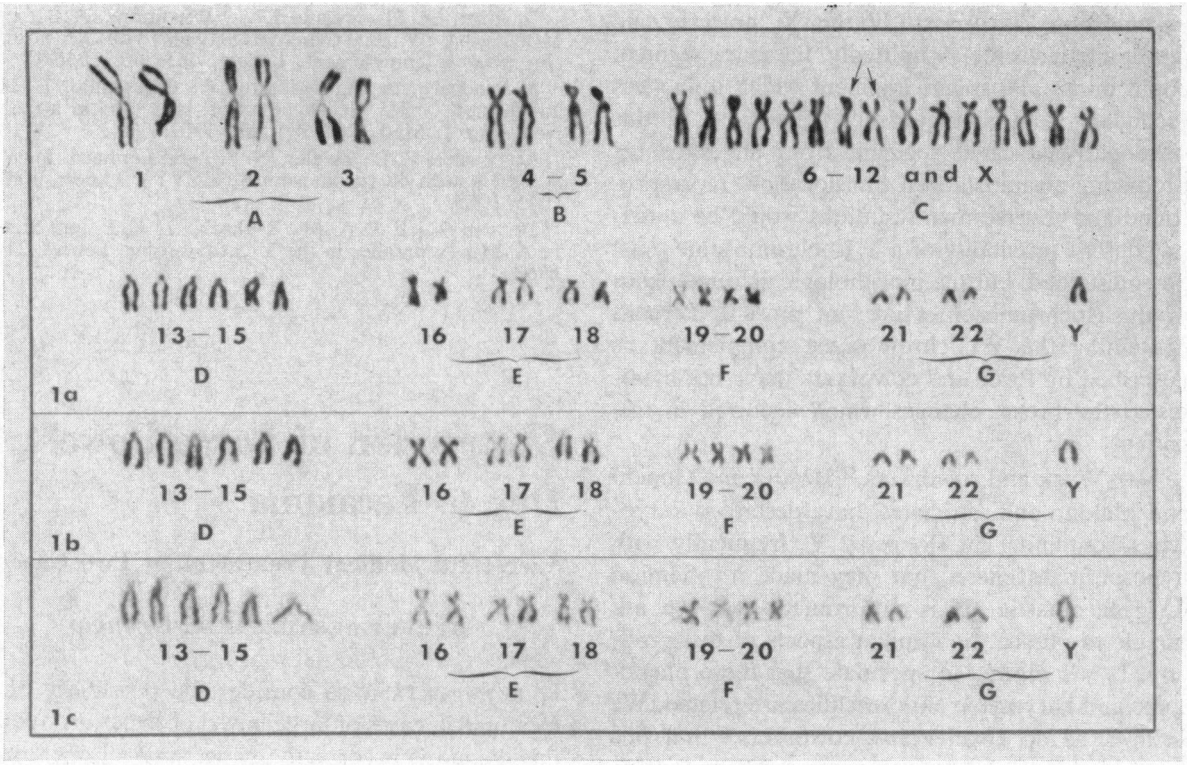

Figure 1.—Karyotype from three mitotic figures, 1a, 1b, and 1c, the latter two figures including only groups D, E, F, and G. Each figure demonstrates the elongated "Y" chromosome. (Note the C′ in figure 1a marked by arrows).

this patient approached the mass of one of the D chromosomes; however, six D chromosomes were present. In addition, all chromosomes of the E, F, and G groups were present and normal.

We and other investigators[2,4,5,6,10] have observed that there is a decided variation from patient to patient in the size of the Y chromosome. In the present case the Y chromosomes were the largest we have seen. Upon determining the Y_2, Y_f and contraction indices (see Table 1) in our patient, we found indices to equal those cited by Cohen and coworkers[4] in their group of patients with elongated Y chromosomes. This is cited not as an indication of abnormality, but only as a means of placing the size of this Y chromosome in perspective.

Unfortunately, the true parents of this young man are not known. He was adopted by his present parents during his infancy. The large acrocentric chromosome must represent more than an elongated Y chromosome. If this were an elongated Y, we could anticipate witnessing the adherence of "stickiness" of the long arms of the Y; this was found infrequently in the figures analyzed. It is tantalizing to speculate that a translocated segment of another chromosome, possibly the X

TABLE 1.—*Chromosome Indexes in Present Case Compared with Those Reported by Cohen and Coworkers*[4]

Variation in Y Chromosome Length		*Y_f Index**		*Y_2 Index**		*Contraction** Index*	
		Mean	*SD*	*Mean*	*SD*	*Mean*	*SD*
Racial groups	Indian	0.88	0.11	0.29	0.05	0.20	0.06
	Japanese	1.00	0.15	0.33	0.06	0.20	0.05
	Negro	0.92	0.09	0.30	0.04	0.20	0.05
	Jew	0.94	0.10	0.29	0.05	0.18	0.07
	Non-Jew	0.86	0.09	0.27	0.04	0.19	0.06
	Present case	1.28	0.17	0.31	0.03	0.16	0.04

*Y Index = Total length of Y chromosomes / Average total length of "Standard Chromosome," i.e., 2,F

**Contraction Index = Average width of short arm #2 / Average length of short arm #2

chromosome, is present in the Y; however, no donor site is visible. Admittedly, the extra segment could be an autosomal fragment which influences secondary sex characteristics. However, if this were a translocated segment from an autosome producing an unbalanced translocation, more profound and diverse manifestations would be anticipated. The possibility of a Y isochromosome must be entertained, but the morphologic characteristics of the isochromosomes are not present. Persons containing the YY chromosome complement as described by Price and coworkers[7] have not manifested the breast changes which occurred in this patient.

Van Wijck and coworkers,[10] Bender and Gooch[2] and Makino and associates[5] have described karyotypes containing an elongated Y, frequently with hypospermatogenesis, but they made no mention of gynecomastia. It is unfortunate that we are unable to pursue the familial aspects of this problem. It is tempting to speculate that these phenotypic and karyotypic abnormalities are related. We believe, as do Bender and coworkers,[2] that one must be cautious about interpretations associating phenotype with chromosomal variations, especially involving the Y chromosome.

Summary

An adult male college student sought medical care because of enlarged breasts. Mastectomy was done and the pathologist's report was consistent with gynecomastia. Testicular biopsy show disorganization of spermatogenesis and hypospermia. A large Y chromosome found in the chromosome karyotype was not accompanied by other cytogenetic abnormalities in the proband. Various possibilities as to the origin of the large Y chromosome were considered.

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Obstruction of Large Bowel Due to Fecaloma

Successful Medical Treatment in Two Cases

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IT IS IMPORTANT to consider the possibility of a fecaloma in cases of large bowel obstruction. When this diagnosis is correctly made, surgical operation can be averted. The following two cases are illustrative.

CASE 1.—A 50-year-old caucasian woman entered UCLA Medical Center for the 16th time on 30 August 1965 with complaint of crampy lower quadrant pain for four days. She had not had a bowel movement in that time. The patient had had numerous abdominal surgical procedures in the past. On examination a left lower quadrant mass was palpable and high pitched sounds were heard on auscultation. The patient was mildly dehydrated but blood and urine examinations were within normal limits. Clinically, the diagnosis of large bowel obstruction was made and a plain film of the abdomen was obtained which showed a dilated transverse and descending colon with large fecal masses in the transverse colon. A barium enema was given and barium would not pass beyond a sigmoid mass despite elevation of the barium container and manual manipulation (Figure 1A). With air outlining the proximal extent of the mass, it was shown to be an elongated sausage-shaped structure (Figure 1B). On this evidence, supported by the presence of the large fecal

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